Why Does My Baby Need More Testing for Alpha Thalassemia?

State law requires that all babies have the newborn screening test before leaving the hospital. A few drops of blood were taken from your baby's heel and tested for certain diseases. Your baby now needs more testing as soon as possible. Not all babies with an initial "positive" result will have an alpha thalassemia disease.

California Department of Health Services

Newborn Screening Program
Genetic Disease Branch
www.dhs.ca.gov/gdb

ED3117

Why Does My Baby Need More Testing?

Your baby's test showed a "positive" result for alpha thalassemia. More testing is needed to find out if your baby has this disease. Not all babies with an initial "positive" result will have this disease.

Babies can look healthy at birth and still have alpha thalassemia. If untreated, this disease can cause severe health problems.

What Is Alpha Thalassemia?

Alpha thalassemia is an inherited condition that affects the special protein called hemoglobin which is in our red blood cells. Red blood cells have an important job. The hemoglobin in red blood cells pick up and carry oxygen from the lungs and take it to every part of the body. A person with alpha thalassemia makes less hemoglobin than most people. This can cause anemia.

How Does a Baby Get This Disease?

A baby with alpha thalassemia inherited a gene from each parent. Both parents must carry the gene for a baby to inherit this disease.

Is Alpha Thalassemia Common?

Alpha thalassemia is more common in people from China, South East Asia, the Philippines, other Asian countries, and Mediterranean and Middle Eastern countries. About 1 in every 10,000 newborns in California (about 51 babies a year) is born with an alpha thalassemia disease. However, anyone can have any of the following types of alpha thalassemia.

Alpha Thalassemia Trait:

People with alpha thalassemia trait can have mild anemia. This causes no serious health problems. However, the red blood cells will be smaller than usual. This is often mistaken for iron deficiency anemia.

Hemoglobin H Disease:

People with hemoglobin H disease have mild to moderate anemia. Sometimes the anemia can get worse and medical attention is needed.

Hemoglobin H-Constant Spring Disease:

Hemoglobin H-Constant Spring disease is a more severe form of hemoglobin H disease. People with hemoglobin H-Constant Spring disease have moderate to severe anemia which requires medical attention and treatment.

What Should I Do Right Now?

- Return to the doctor's office or hospital lab as instructed for more testing for your baby. The doctor will also ask one or both parents to be tested.
- Follow the doctor's advise on treatment. This will probably include taking your baby to a pediatric hematologist and/or sickle cell disease special care center.
- Your child should avoid coming into contact with mothballs and fava beans. Both of these can cause severe anemia in someone with alpha thalassemia.

How Will I Know My Baby's Results?

The Newborn Screening staff reports the result to the doctor. The baby's doctor will let you know the results.

How Are These Diseases Treated?

Most people with hemoglobin H disease can lead relatively normal lives with proper treatment. Children with hemoglobin H disease should get prompt medical attention when they become ill since viral infections and fever can cause the red blood cells to break down faster. In addition, certain household products (such as mothballs), medications, and fava beans can cause severe anemia if a child with hemoglobin H disease comes into contact with these items.

Where Can I Get More Information?

Call the doctor or the Newborn Screening staff listed below for any questions or concerns about hemoglobin H or hemoglobin H-Constant Spring disease confirmatory testing.